Jorge Sequeiros

Personal Details	
Name	Jorge Sequeiros
Dates	1952
Place of Birth	Porto
Main work places	Porto
Principal field of work	Neurogenetics

Interview	
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INTERVIEW WITH PROFESSOR JORGE SEQUEIROS, 15 OCTOBER 2011

PH = Interviewer (Peter Harper)

JS = Jorge Sequeiros

PH It's Saturday, 15th October 2011, and I'm talking with Professor Jorge Sequeiros from Portugal at the International Human Genetics Congress in Montreal. Jorge, can I start at the beginning and ask, when were you born and where?

JS I was born in 1952. Well, I'm a kind of inbred strain because I was born in Porto, I lived most of my life in Porto, except for the three years at Hopkins, and I work and live in Porto again, so…

PH And did you grow up and go to school in Porto?

JS Yes, everything in Porto (grammar, secondary and medical school), except the three years at Hopkins. And of course some minor periods in Paris with Arnold Munnich, during four years, and some time in Montreal, here at McGill, for brief periods, where I begun trained my first PhD students.

PH May I ask, did your family have any particular medical or scientific background at all?

JS No, not at all.

PH What did your parents do?

JS My father came from what we think was an aristocratic family; bankrupt [laughter] from the north of the country (Ponte do Lima, Minho). Apparently the origins were somewhere in Galicia; someone came from Galicia, and the pedigree is documented up to 11th century, so before the nationality in Portugal began (1140). And he was the founder of the Portuguese Sequeiros: all the Sequeiros in Portugal are related

PH Do you know that from Y chromosome studies or just from -

JS No, just from history. Only from history. And then now, with internet, I find Sequeiros all over the place because it's basically a Spanish, a Galician, name and so it got spread all over South America and so on. I find football players, artists, all sorts of people [laughs], when I try to look sometimes for articles I wrote and things I did and these guys pop up like that. Recently, I even gave a lecture in Lima, Peru, where there as a neurologist in the audience named Sequeiros.

PH So, a distinguished family.

JS Yeah, it was earlier a distinguished family, because my grandfather, who apparently was a dandy, never worked in his life. I never knew him because he was much, much older than my very beautiful grandmother, whom I only know from family pictures. And so he died and left her with many children: they had 15 children, altogether, though some of them deceased very early; when he died he was completely broke and most of the remaining children had to emigrate. So, a couple of them went to Brazil, some to Mozambique, my father went to Porto, which was the nearest big town. And so my father began by being a message boy, in local commerce (Porto was then a thriving commerce town), and went up to become a manager and then a partner of commerce, and thus became basically a merchant. My mother came from a very humble family; they were very humble, from the outskirts of Porto and they married very, very young; they met when she was 19, he was 22; they married a couple of years later and they lived in Porto ever since, so I was born there and everything started there for me.

PH Were there any particular factors you can think of that brought you into medicine?

JS Into medicine? Well, I have posed that question myself, several times, but I think I always have the feeling of wanting to be helpful, of wanting to do something for the community and I thought medicine was a great way to do it. But, when I think of very specific facts, I always remember, when I was adolescent, pre-adolescent, 10, 11, 12, there was a series of biographies, very, very popular in Portugal then that I read most of them. And I loved reading these biographies, but the ones I remember that marked me the most were the ones of Louis Pasteur, Marie Curie and then Albert Schweitzer and Florence Nightingale. These were the 4 that I remember better, and then there was Geronimo, the apache leader, in addition to Mozart, Beethoven, Edison, and many others. So of all I read, while still very young, and I read many, those 5 were I think the ones that marked me the most… I don't think really there was anything else in particular that may have triggered. Well, perhaps our family doctor (Pais Clemente), who was an extremely nice and kind person, who paid us frequent house calls, and treated my from an acute meningitis when I was 6.

PH So you went to school in Porto?

JS In Porto.

PH And what year did you start in medical school?

JS I started in 69 and finished in 75. I was the youngest of my class. It's very difficult to explain, but it has to do with my birth date being by the end of October. Because classes began in early October, you had to be a certain age before that date to enter grammar school. So I was able to take the entrance exam (my birthday had to be before the 31st December) to enter the lyceum, secondary school, though I was not old enough to take the graduation exam to finish grammar school, because of an odd legislation lag. And it was very important for my parents that I went in as young as possible. I'm not so sure that was a good thing. [laughs] But I had to live all my life with the burden (mixed with the pride!) of always being the youngest one in my cohort.

PH Were there any particular parts of your course that for you were outstanding?

JS Yes, of course. I remember the parts and teachers I had in pathology, I had very very good teacher Daniel Serrão, an extremely conservative guy [laugh], but he was an excellent teacher. I remember he introduced me to immunology, at the time the the distinction between B and T lymphocytes were coming out, as well as the higher incidence of scrotal cancer in chimney cleaners in the UK. I still remember well some of his classes, very vivid classes and very updated, and very interesting. I think that was greatly responsible for me getting the pleasure and willing to teach, to become a teacher. And then, the crutial momnt for my carrer was in the third year of medical school, when I had medical genetics. It was not still called medical genetics, but a part of general pathology. Half was immunology, the other half was genetics. This was the first course in genetics in the whole country, it was '71.The professor was Amândio Tavares; he offered us, the students, the opportunity to do research in his lab; this, for undergraduate students, at that time, was completely unheard of. So I immediately grabbed that opportunity, because I always had the idea that I was very much willing to do research. That's how I came into genetics at that time, at the 3rd year at medical shool.

That kind of research was basically applied cytogenetics: the clastogenic properties of some common chemicals. I remember we were "breaking" chromosomes with caffeine [laughs], theophylline, with lithium; with mescaline, with LSD. We had all those experiments and actually came across what later were called the fragile loci. I have a publication (in a Portuguese medical journal) stating where I show that those chromosomal breaks are not at random. We still didn't have band karyotyping at that time, so the location of the breaks were more or less in a given point, but it was enough to see that they were not at random.

PH Was that on human blood then?

JS That was on human blood. Yes, first in vitro, but later on the studies we did were the same but with the bipolar ("maniac-depressive") patients the blood of whom I was "collecting from different hospitals for the study of chromosomal breakage and lithium level dosage..This was in the early 70s.

PH That must have been very unusual really -

JS It was unusual -

PH For an undergraduate course to be studying human chromosomes.

JS Yes, it was. It really was. And that was available there; nowhere else in the country. So, I just happened to be in the right place and at the right time, and have Amândio Tavares as a medical genetics teacher, to have that opportunity, and then…

PH So then you qualified in which year?

JS In 75.

PH 75. And what did you do next?

JS Well, just let me go a little bit before that, because I think this is important.

PH Sure.

JS So with this, with this research work, as a third year medical student I'd already got a few publications, and I went to the first scientific meeting to make my first oral presentation in 1972. So that's where I mark the beginning of my scientific career. 72, in Cordoba, Spain. At the time there was still no Portuguese Society of Genetics, but there were Portuguese/Spanish joint meetings, every year; once in Portugal, then in Spain. A little biased, because it was easier for us than for the Spanish [laughs], having a much lighter country. So that's where it began. And then I, and in my graduation, in 75, the Portuguese revolution was had been in 74, just one year-and-half before I graduated. Beforehand, I was using my summer holidays to travel all around Europe using my scholarship money; my parents were not rich, but I had a scholarship from the Gulbenkian Foundation and every year they saved that money for me. And with that money I was able to pay a plane trip to somewhere, Scandinavia, Holland and so on. And we have very long holidays, like two and a half or even three months summer holidays at that time; not any more…

So I used those holidays to go and work for some time. I've been to Hastings, picking hops and to Cambridge, picking up apples; then in Amsterdam, working in interior refurbishment of old houses; I worked at the Forest hotel in Lidingö iStockholm, where the Rolling Stones were at the time, where I met them. [laughter]

And of the reasons I travelled in Europe for the summer was also to look for a place to exile myself, as we has the war in the old colonies, which I refused to join. As a university student, at that time, we have a postponing until we graduated, as the regime wanted officers, for the army. So, if you didn't fail any year and if you did well at university, you'd be able to finish your degree, but then you would be incorporated in the army. So, if there was no revolution in the meantime, April of 74, my choice had been taken already: to exile in Amsterdam or Stockholm or Paris, I was still undecided which of these three places I would elect.

So I was also using my summer holidays, as a survey for that. But, luckily, April 74 came, and this was not needed anymore. But, at the same time, came another thing: a very political and a civil engagement. As students and young doctors, we did literacy campaigns to help countryside people learn to read; so there were campaigns throughout the country, and we started also with a few colleagues, very spontaneously, doing medical clinics in distant parts of the country, volunteering our services in places where they had no doctors at all. And, actually this became a practice; it was called the "periphery" year, where after a medical internship of two years, after graduating from medical

school, we had a third year of civil service at the most peripheric and in need regions of the country. Thus I spent a year in the mountains in the northeast of Portugal, going from village to village, and doing weekly 24h emergency care for a vast region and population, all by myself or just with another colleague.

And that was one of the most wonderful years I had in my life and gave me very much the willing to do clinical medicine and not so much research. And, much to Amândio Tavares' dismay, I decided to leave the university and dedicate myself to medicine at Santo António's hospital. And so then I entered the speciality of internal medicine, which I completed and I did my training at Hopkins, 82 to 85.

PH How did that come about? How did you make the link with Johns Hopkins?

JS Well, this was Corino de Andrade, the neurologist who described familial amyloid polyneuropathy, and published it in Brain, in 1952, the year I was born. He was one of those people with a great vision and really able to gather people around him from different things. And, when I was doing an optional rotation in neurology, in 76, at his service, exactly at the time he retired, but was still very active and present for the rounds and seminars, etc. After my "periphery year" I returned to his service. "Ah, you must go. You have genetics experience; we have these pedigrees (of familial amyloid neuropathy and the then recently discovered Machado-Joseh disease)…" He had this nasal voice. "So you have to come and work with us. And the person to begin to do that with is Victor McKusick, at Johns Hopkins, and I'm going to write him a letter" and so on. So he wrote one letter, two letters, but no answer came. But then there was International Congress of Human Genetics, in Jerusalem, which I think was in 1981 -

PH About then.

JS Yeah, September 1981. So he wrote a third letter that I delivered in hand to Victor himself. I handed it to him, and he said, "Well, just make your preparations and come when ever you can." I did the ECFMG examinations, and TOEFL, and in August 82 there I was in Baltimore.

PH How had Andrade come to know McKusick?

JS He didn't know him personally. I don't think he ever met him, except when McKusick went to visit, by invitation, to Porto (at the time I was in Baltimore); it was the first time they met. But he knew everything and knew everyone. And he was a very, very attentive person; he was very much aware of what was happening in every field. And a well known scientist all over the world.

PH So when you got to Hopkins were you, at that time, can I ask, were you on your own or were you married by this stage?

JS I was married and I had already my two daughters. Sara was 2 years and Elsa was only 2 months. So there I was with two very very young children -

PH And you all went to Baltimore?

JS We all four went to Baltimore. I went ahead to find a home, to install myself and they joined me a couple of months later.

PH Yes. And what was -

JS - not easy, of course.

PH No. We had a very similar experience.

JS I had a very, very small scholarship; at a time our national currency was going down and the dollar was going up. In one year my scholarship was reduced to half. So, there you have it. But that's another story.

PH What was the main work then that you did when you were at Hopkins?

JS Well, the idea, the basic idea at the very beginning, was to do with something for my PhD. And my PhD was around Machado–Joseph disease, because Corino Andrade had already been to the Azores in 76, January 76, so that's when the whole story of Machado, the Machado family and the Joseph family came about and disease was unified; everything was put together. And he visited the Azores in 76 with Paula Coutinho, so that's at the time I entered the Santo António's hospital and met him and all this meant. It was more or less decided that I was going to do my PhD thesis on Machado–Joseph disease. Corino Andrade also was a founder and president of the Scientific Council of a new medical school, ICBAS, associated to Stº António's Hospital, where I began teaching medical genetics.

There were all these Portuguese-American communities in the States, but not in Baltimore. So I had to travel back and forth to New England and the Boston area and to San Francisco, mostly the Oakland area, and I met the Machados and the Josephs received a lot of help from the International Joseph Diseases Foundation, led by an extraordinary woman, Mrs Rosemary Silva, from Livermore, California. She started this foundation and was holding family meetings and clinical sessions. Patrick McLeod came along, Roger Rosenberg, and a bunch of neurologists, geneticists, genetic counsellors, and we had like one week, one full week each time, seeing patients, drawing and updating pedigrees and doing field work in both the coasts of the US. In fact, this was my hobby.

This was supposed to be my major aim, but actually it was my hobby. It was done during my free time, because at Hopkins I was just a postdoc fellow and I did everything else: I saw patients in clinics, as you know, all those patients with Marfan and Ehlers-Danlos, which were 80% at that time [laughs] and other connective tissue diseases. And I went with Victor who took me in many visits for the LPA clinics, to the Amish country, on various meetings, etc. And that's how I spent my time, that's why my PhD was very much delayed, because it was only when I got back to Portugal, in 85, that I was able to begin treating and begin working on the thesis that I completed in 89. And so, four years later, I defended it in January of 1990, after having returned to Santo António's Hospital (where I still had to finish my residency in internal medicine) and my teaching at ICBAS.

PH So after you'd come back, you had three years in Baltimore, or was it two?

JS Three full years. There's another important thing is that Andrade, Corino de Andrade, together with Amândio Tavares and a few other people that were very visionary in terms of, they wanted to build a new biomedical sciences institute in Porto that would have basic teaching in biology for the first few years, and then you could choose going either to medicine or to biology or biochemistry or something else. Well, that never happened because the government didn't allow it. It was far too advanced to the time, so it eventually became a school with a degree in medicine, a degree in sea life sciences, veterinary and biochemistry, all very much independent but that we tried to teach together the first few years. And he co-opted me to go to that new institute that was being formed and so I began there basically with medical genetics in that institute from scratch.

PH Right, so what was the pattern of medical genetics over the years from where you began? When you started, was it just yourself or did you have a laboratory of any kind?

JS I (re)started at ICBAS, the Institute of Biomedical Sciences Abel Salazar (this was not the dictator; Abel Salazar was a histologist and artist and painter, who had been expelled from the School of Medicine by his homonymous, the dictator. There was some basic cytogenetics, and a molecular genetics lab, all independent from each other).

PH Was that new or had it been in existence from before?

JS No, no, it was new. The whole institute was created in 75, and began in 78-79. I think it opened in 78, while I was still doing my year at the periphery. When I entered in 79. there was basically a cytogenetics lab, run by an agronomist engineer, Tristão Mello Sampayo, there was population genetics (with a "fly room"), led Luís Sieuve Corino Monteiro, who was a veterinarian by formation and there was a molecular genetics lab, that had been begun by Luís Archer, a Jesuit priest and biologist, who trained in

the USA, and introduced molecular genetics in Portugal. So there were these three labs, and they felt that my place would be better in the cytogenetics one, so that's how I entered that group. It was a very different situation and structure from the traditional medical school.

PH So did you have, from the beginning, links with the hospitals that allowed you do to clinics and things like that?

JS Well, I was still finishing, at the same time, my internal medicine residency at Santo António's Hospital, which is just across the street and became actually the teaching hospital for the biomedical sciences institute. The biomedical sciences institute provides the basic years, the three first, and then all the clinical teaching is done at the hospital where Andrade was and where I was doing my residency.

PH This is before you went to Hopkins?

JS Yes, this was before. From 1979 to 1982, though I then returned in 1985.

PH Okay.

JS When I went to Hopkins I already had experience and some training; well, not formal training in clinical genetics, but some experience. The very beginning was with Andrade in neurology. This is also an important fact, a very important factor, all these coincidences which make up your life I [laughs], you know. That was a time of great political turmoil, where governments succeeded one after another. So, when I came back from the year in the periphery, the rule was that we should go back (until we were waiting for the national exam to enter the speciality to the last service in which you were before - and that was neurology. So I eventually spent 3 years in neurology [laughs] while waiting for the exam... When I finally entered the residency in internal medicine, I already had 7 years after I graduated. But it certainly gave me a lot of experience and the opportunity to be in neurology for 3 years and to get linked to the biomedical sciences institute, virtually from its start.

PH So when you came back from Hopkins were you then able to develop a clinical genetics service in terms of clinics?

JS No, I did not. I tried to do that beforehand, while I was still doing my internship and then my residency in internal medicine. During my internship, I had a clinical genetics consultation, at the Department of Gynaecology, due to the willing and understanding of another great men and physician, Albino Aroso (who started family planning in Portugal).

PH Okay.

JS But that was as far as I went and I did it for some time, but I never succeeded in making a genetics service, and then I left to Hopkins, so I didn't have much time; I only had 2 years. So this was from 80 to 82. In 85, when I came back, I still had my thesis to write and I had all that, so I pursued my "clandestine" clinical genetics consultations. Then, during the rest of my residency, I managed to convince my director (Antunes de Azevedo) to dedicate one of our outpatient clinic periods to clinical genetics. So, as I was seeing patients, internal medicine patients, … when they were dismissed from the clinic, I began filling the consultation gradually with genetics patients. But, later on, when I finished my residency, I left the hospital to go full time to the university, and that was a choice I had to make. And it's still something I feel like a failure not having been able to build a medical genetics service in the hospital, but you know, I was still too young to be taken seriously, at that time. I was not at all experienced with the politics and policies of the hospital and I wasn't able, at that time, to overcome all the bureaucratic obstacles from inside and outside the hospital.

PH Was there any traditional of joint academic hospital appointments? Or were the two institutions really very separate?

JS They were just separated by a narrow street but [laughter] -

PH A very wide, narrow street.

JS Yes, the gap was enormous, and still is. They belong to two different ministries; there is some kind of protocol, but this works only one way, so that the physicians from the hospital may have university appointments, but we physicians that have opted for the academic career cannot have hospital appointments. I tried it several times, with different people, but was never able to revert it.

So eventually, a crucial thing happened. In 1992, sorry I am getting much more advanced now…; in 1992, soon after we joined the European Union, with European money, there was this big, big project to create new research institutes. So, I was proposed to create a new research group (UnIGENe – Unit for Genetics and Epidemiological Investigation of Neurological Diseases), which I am still leading, at IBMC, the Institute for Molecular and Cell Biology. My situation is that my full salary comes from the university; I teach at the Biomedical Sciences Institute, but my physical place is at IBMC where I jhave my office, my research group, grants, projects and so on. And, in that research institute, I eventually managed to create a genetics centre, with an outpatient clinic and a molecular genetic testing lab; which is very unusual at a research institute. What I could manage to do at the hospital, what I could not do at ICBAS, I was able to at IBMC. So, in addition to the neurogenetics research lab (created at IBMC in 92, though could move in only in 97), we created an outpatient clinic and diagnostic lab, and so I'm running all three now at the same time, and giving classes, well, having 4 or 5 jobs and being paid for one, basically. [laughs]

PH When you came back after your time in America and you finished your thesis on Machado-Joseph, did you then start to broaden your area of interest, and was that still more in neurogenetics? Or did you try to go outside neurogenetics at all?

JS No, I deliberately chose not to go outside neurogenetics, because I wanted to focus very much in neurogenetics and particularly on the repeat diseases. We began with Machado-Joseph and the other ataxias came along (dominant and recessive), because you cannot do one without doing the others, and then we had Huntington which had been the paradigm for all these diseases. So, with Huntington, the funny thing was that we began delivering services because no one was offering testing for Huntington in the country. So now we are doing some research in HD as well. It was exactly the other way around with Machado-Joseph: when, at a given point, we felt that we had been asking so much to those families for such a long time, that now that we had a gene and mutation we could offer testing, it was time to offer them something. And we began to offer presymptomatic testing and the genetic testing in general, and counselling following the international guidelines for Huntington in 98. And so that's how it was. So it's still very much centred in neurogenetics, both in research and in services to the community.

PH Yes. Can I ask, during this time, what had been happening to medical genetics in Portugal as a whole? Because from what you say, at the time when you went to America really, there was not so much. Perhaps you can give me an idea about how things have evolved more generally and maybe it's best for you to go right back to as far as you can.

JS That is in the 50s, as I was mentioning earlier on. The first teaching of genetic was in Coimbra, by Aurélio Quintanilha, the father of Alexandre Quintanilha, the director of IBMC. He was a botanist and basically, at the time, it all began after the World War and the crisis with the agricultural policy. Aurélio Quintanilha was very politically active against Salazar and after some time eventually was exiled. But there was this other guy Tamagnini, who followed exactly the same agricultural politics of Mussolini. As far as I know, these are the first two people who actually did something we can call genetics, in Portugal. Quintanilha formed a few people, one of them being Serra, who became well known. He was not very politically involved, but he had to make a stand at one time and he also was sent to exile, as happened with many well known people at that time. Those are what whom we can call the very early geneticists in Portugal, doing agricultural and veterinary genetics.

PH Had they introduced cytogenetics in the places they worked? Because much of cytogenetics started off in botany and agriculture.

JS That's what happened exactly. They did cytogenetics of wheat. Miguel Mota and Tristão Mello Sampayo, The first human karyotype was done by Amândio Tavares in 1953, still with a number of chromosomes uncertain. He, himself, dates the beginning of human genetics and medical genetics in Portugal at that moment, in 1953. There were two other major events: Amândio Tavares was asked to study a teratoma from a surgeon colleague, and he did a review on teratomas that he published in Lancet in 1955; he also did a study on sex chromatin published in the Journal of Pathology and Bacteriology, and, as a result, he got an invitation to London, to a meeting organised by CIBA, where he met Paul Polani and Anders, who became his main connection. As he tells, they were expecting someone in their 50s, while he was only in his early 30s at the time. When he came back from that meeting, he began doing human karyotyping, … oh, and he mentions also that, at the meeting, there were already some rumours that Tjio had found the right number of chromosomes, so he began doing tissue culturing and karyotyping and, as a consequence, doing clinical cytogenetics, which led him to reach a major interest later in genetic counselling.

PH Now where… was this all in Porto?

JS This was all in Porto, at the Faculty of Medicine; this was all in the 50s.

PH Was there anything happening in Lisbon or in other parts of Portugal?

JS Not in human genetics, no. Only at the Institute of Agriculture and then the Gulbenkian Institute, where Luís Archer was. Archer was a biologist, then he decided to become a priest, a Jesuit priest, and he went to the States, where he formed in molecular genetics. So, when he came back, he wanted to go back to do only theology; but being in a religious order, he had to do what he was told to do [laughs] and his superiors said, "No way! You have to go back to science, because we need people there. You've trained as a scientist, so you have to go back." So, much against his will, because he wanted to dedicate himself to theology and social sciences, in general, he was forced to create molecular genetics in Portugal, which he did, in 1971, in Lisbon at the Gulbenkian Institute in Oeiras, near Lisbon; and he also helped molecular genetics in Porto, at the Biomedical and Genetics Institute.

PH Do you think there was much influence from Spain in early genetics?

JS No, it was the other way around. Amândio Tavares, who is married to a Spanish lady, he influenced a lot the beginning of genetics in Spain. People like Goyanes, in Galicia (he's a godfather to a child of Goyanes), they are very close friends, and with Izquierdo and Egozcue. Together, they published book in Spanish that was used by medical students both in Portugal and Spain. But it was Amândio Tavares who the one who influenced very much the beginning of it all, both in Portugal and Spain. And, as I mentioned earlier, these Portuguese-Spanish joint conferences that took place annually and which were very important to get them together. They were a considerable scientific meeting and a true school in genetics for many of us.

PH How did medical genetics develop from the time that you had established something in Porto, then how did it come together and start to develop over the country?

JS Well, again it was Amândio Tavares who began training people. He was the trainer of Heloísa Santos, still in Lisbon, though she was originally from Porto and of Maria de Jesus Feijóo, who died a few years ago. Both were the founders of the two medical genetics services in Lisbon. There was also Ferraz Júnior, a paediatrician from Lisbon, who did not train with Amândio Tavares, though he had some proximity with him. Then you have Jacinto de Magalhães, who created the Medical Genetics Institute (IGM) in Porto. He also trained with Amândio Tavares,,actually at the same time I did, only that I was a young student and he was a established paediatrician by then. Jacinto de Magalhães also had a short stay with Jean Frézal, and later started a series of conferences in Porto, inviting mostly specilaists from

France and the UK. These conferences of the IGM became a very important school of medical genetics for all of us as well.

Amândio Tavares is very simple and humble and always kept a very low profile. But this was not enough for Jacinto, who had other ambitions. Jacinto left and founded what later became the Institute of Medical Genetics, in Porto. Because Amândio Tavares, with his teaching appointment, never succeeded in creating a clinical genetics service in the São João Hospital (and both the teaching hospital and school of medicine are in the same building, there isn't even a street separating them…). Though he performed genetic counselling and saw genetics patients in genetics service he created at the School of Medicine. So we have this particularity of having two schools of medicine in the same university, in Porto: the old one, where I graduated and the only one until 75; and the one Corino de Andrade later created with other people, and where I am teaching, the Biomedical Sciences Institutes (ICBAS). But I was, I lost track of what I was saying…

PH We were talking about the different, the development of medical genetics overall, but can I just, on that point of the two units and medical schools in Porto; has it been possible for them to come together at all or are they still very separate?

JS They are still very separate. I think this will eventually be resolved, but they are still very separate; as widely as they can, though some of us have managed to make joint events and even courses. Because, what I was saying earlier that I forgot, was that Amândio Tavares himself, was a full professor at the University Faculty of Medicine, not in the University hospital; his appointment was with university and he was never able to create a medical genetics service in the hospital, as it was his wish long before his retirement. A medical genetics service was created in th São João Hospital just 2 years ago.

PH And the other medical genetics unit then, is this more paediatrically orientated or -

JS Yes, IGM, the medical genetics institute created by Jacinto Magalhães, actually grew from a genetics service of a paediatrics hospital, but later developed other services and became a large centre. So, basically we have the two main central and teaching hospitals in Porto. São João, that just created a small genetics services 2 years ago; and Santo António, where I was, that never did. Now it's not independent anymore, because the National Health Institute, in Lisbon, which has also a large genetics lab, just took over IGM, in Porto. But right now, the most likely scenario is that the clinical geneticists are most probably going to Santo Antonio hospital, which will finally will have its own medical genetics service, after all. Well, sometimes things are like that.

PH What about your association because, well, I remember, coming to one of the meetings myself that you have a Portuguese Association? It was the founding meeting. Tell me how that came into being.

JS The founding meeting was in 1996. Both you and Rodney Harris were invited speakers. Before, we had only a Portuguese Society of Genetics, created in 1973, that was very much dominated by the plant and animal genetics people, much as a consequence of the Portuguese-Spanish conferences that took place then. At a certain time, some of us we felt the need to create a separate Human Genetics Society; this was really a nucleus of people, that included Heloísa Santos, Carolino Monteiro, Isabel Marques Carreira, Sérgio Castedo, Purificação Tavares and myself, that created the Portuguese Society of Human Genetics and this was the very first meeting to which you and Rodney Harris were the invited speakers, and this happened in 6 December of 96. So that's when Portuguese Society for Human Genetics began., and it is still in good shape and growing.

PH Good.

JS Heloísa was the first president; I was the president-elect. I wrote the statutes; we followed the same structure as the European Society of Human Genetics and that's how we have been living since then.

PH Are there other things that you'd like to tell me about either your development of your own work -

JS My genetic speciality?

PH Or things in general; anything which I haven't asked about that you'd like to put on record.

JS Yes, I think the most important thing that we have to talk about besides the creation of these two societies, is the creation of the specialty of clinica genetics and the medical genetics college. Since the 70s, there was something called Competência in medical genetics,inside the Portuguese Medical Association, that certifies the clinical specialties. This so called "competence" was something even below a subspecialty,, that you could acquire after having a primary specialty. And this went on for long, long years. Its first president was Jacinto de Magalhães, then it was Amândio Tavares and the third one was myself. And after we all struggled for many, many years, we finally succeeded, on 2 April 1999, in creating the Medical Genetics specialty.

Still in 1999, we began visiting and certifying the medical genetics services and in 2000 we begun the admissions by consensus of those people who were already qualified by experience and having trained elsewhere and formed the Medical Genetics College. In 2002, the first residency program began; so the first cohort of 5 students, fully trained in Portugal, came out in 2007, and we are now about 45 certified medical geneticists in Portugal, which does not mean that all of them are active in the National Health Service. Some of them are already retired; some are still exercising their primary speciality, not medical genetics; and others are primarily in teaching or research. So we have something like 25 to 30 medical geneticists doing clinical work, in 5 medical genetics services in the country. So we have 2 in Lisbon, 2 in Porto and 1 in Coimbra, which is not too bad for the needs of the country, though some of these are small and would need to be further developed and staffed.

PH And your population of Portugal is -

JS Ten million.

PH Ten million.

JS We have another major development and this was a promise I have made of myself after being in United States and working with genetic counsellors, and I just told Barbara Bernhard exactly that the other night, was that after we managed to succeed creating the medical specialty, we had to create a master's course for genetic counsellors, which I began at the university, at the Institute for Biomedical Sciences two years ago, and the first cohort is coming out in just two weeks. [laughs]

PH Good.

JS The first 6 genetic counsellors. Milena Paneque, a genetic counsellor who was trained in Cuba, has been of much help and crucial in running this master course with me, for the last two years. And the first three tare coming out now, on the 27th October, and the other three in December. So the first cohort will be completed then.

PH Good.

JS Step by step we'll be there.

PH Well, many thanks Jorge. I will finish there and it's good of you to spare the time and I'll turn the machine off, but many thanks.